



## COL4A5 gene

collagen type IV alpha 5 chain

### Normal Function

The COL4A5 gene provides instructions for making one component of type IV collagen, which is a flexible protein. Specifically, this gene makes the alpha5(IV) chain of type IV collagen. This chain combines with two other types of alpha (IV) chains (the alpha3 and alpha4 chains) to make a complete type IV collagen molecule. Type IV collagen molecules attach to each other to form complex protein networks. These networks make up a large portion of basement membranes, which are thin sheet-like structures that separate and support cells in many tissues. Type IV collagen alpha3-4-5 networks play an especially important role in the basement membranes of the kidney, inner ear, and eye.

### Health Conditions Related to Genetic Changes

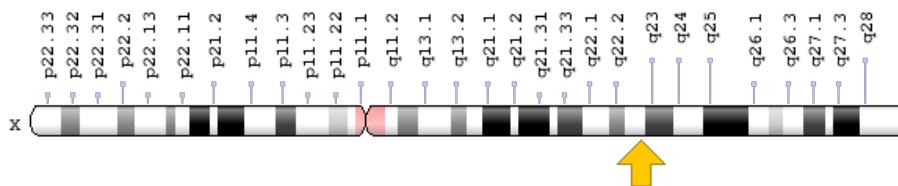
#### Alport syndrome

More than 400 mutations in the COL4A5 gene have been found to cause Alport syndrome. Most of these mutations change single protein building blocks (amino acids) in a region where the alpha5(IV) collagen chain combines with other type IV collagen chains. Other mutations in the COL4A5 gene severely decrease or prevent the production of alpha5(IV) chains. As a result, there is a serious deficiency of the type IV collagen alpha3-4-5 network in the basement membranes of the kidney, inner ear, and eye. In the kidney, other types of collagen accumulate in the basement membranes, eventually leading to scarring of the kidneys and kidney failure. Mutations in this gene can also lead to abnormal function in the inner ear, resulting in hearing loss.

## Chromosomal Location

Cytogenetic Location: Xq22.3, which is the long (q) arm of the X chromosome at position 22.3

Molecular Location: base pairs 108,439,844 to 108,697,545 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- ASLN
- ATS
- CA54
- CO4A5\_HUMAN
- collagen IV, alpha-5 polypeptide
- collagen of basement membrane, alpha-5 chain
- collagen type IV alpha 5
- collagen, type IV, alpha 5
- collagen, type IV, alpha 5 (Alport syndrome)

## Additional Information & Resources

### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): A model of the molecular structure of a basal lamina  
<https://www.ncbi.nlm.nih.gov/books/NBK26810/?rendertype=figure&id=A3581>
- Molecular Biology of the Cell (fourth edition, 2002): Basal Laminae Perform Diverse Functions  
<https://www.ncbi.nlm.nih.gov/books/NBK26810/#A3583>

## GeneReviews

- Alport Syndrome and Thin Basement Membrane Nephropathy  
<https://www.ncbi.nlm.nih.gov/books/NBK1207>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28COL4A5%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

## OMIM

- COLLAGEN, TYPE IV, ALPHA-5  
<http://omim.org/entry/303630>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_COL4A5.html](http://atlasgeneticsoncology.org/Genes/GC_COL4A5.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=COL4A5%5Bgene%5D>
- HGNC Gene Family: Collagens  
<http://www.genenames.org/cgi-bin/genefamilies/set/490>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=2207](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2207)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/1287>
- UniProt  
<http://www.uniprot.org/uniprot/P29400>

## **Sources for This Summary**

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- GeneReview: Alport Syndrome and Thin Basement Membrane Nephropathy  
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*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17396119>

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